

Client: Example Client ABC123
123 Test Drive
Salt Lake City, UT 84108
UNITED STATES

Physician: Doctor, Example

Patient: Patient, Example

DOB 1/20/2001

Gender: Female

Patient Identifiers: 01234567890ABCD, 012345

Visit Number (FIN): 01234567890ABCD

Collection Date: 00/00/0000 00:00

Hemophilia A (F8) 2 Inversions, Fetal

ARUP test code 2001755

Maternal Contamination Study Fetal Spec

Fetal Cells

Single fetal genotype present; no maternal cells present. Fetal and maternal samples were tested using STR markers to rule out maternal cell contamination.

INTERPRETIVE INFORMATION: Maternal Cell Contamination, Fetal Specimen
Please refer to fetal report for interpretation.

Maternal Contam Study, Maternal Spec

whole Blood

For quality assurance purposes, ARUP Laboratories will confirm the above result at no charge following delivery. Order Confirmation of Fetal Testing and include a copy of the original fetal report (or the mother's name and date of birth) with the test submission. Please contact an ARUP genetic counselor at (800) 242-2787 extension 2141 prior to specimen submission.

F8 INV FE Specimen

Cultured Amnio

Hemophilia A (F8) Inversions Interp

Negative

H=High, L=Low, *=Abnormal, C=Critical

Section 79-1 of New York State Civil Rights Law requires informed consent be obtained from patients (or their legal guardians) prior to pursuing genetic testing. These forms must be kept on file by the ordering physician. Consent forms for genetic testing are available at www.aruplab.com. Incidental findings are not reported unless clinically significant but are available upon request.

Negative for the familial inversion in the F8 gene.

INTERPRETATION

According to information provided to ARUP, the mother of this fetus is a carrier of the factor 8 (F8) intron 22-A inversion. Neither the familial F8 intron 22-A inversion nor the intron 1 inversion were detected in this prenatal sample; therefore, this fetus is predicted to be unaffected with hemophilia A.

RECOMMENDATIONS

Genetic consultation is recommended. For quality assurance purposes, ARUP Laboratories will confirm the above result at no charge following delivery. Order Confirmation of Fetal Testing and include a copy of the original fetal report (or the mother's name and date of birth) with the test submission. Please contact an ARUP genetic counselor at (800) 242-2787 extension 2141 prior to specimen submission.

COMMENTS

Reference Sequence: GenBank # NM_000132.3

Note: A positive familial control was tested.

This result has been reviewed and approved by Rong Mao, M.D.

BACKGROUND INFORMATION: Hemophilia A (F8) 2 Inversions

CHARACTERISTICS: Severe deficiency of factor VIII clotting activity leading to spontaneous joint or deep muscle bleeding. Moderate to mild deficiency is associated with prolonged bleeding after tooth extractions, surgery, or injuries and recurrent or delayed wound healing.

INCIDENCE: 1 in 4,000-5,000 live male births worldwide, rare in females.

INHERITANCE: X-linked recessive. Of simplex cases, 85 percent of mothers are carriers and 10-15 percent of boys have a de novo mutation.

PENETRANCE: 100 percent in males and 10 percent in females.

CAUSE: Deleterious F8 gene mutations.

CLINICAL SENSITIVITY: 51 percent of mutations causing severe hemophilia A are detected by F8 inversion testing. This assay does not detect F8 mutations associated with mild or moderate hemophilia A in males.

METHODOLOGY: Intron 22-A and intron 1 inversions detected by inverse PCR and electrophoresis.

ANALYTICAL SENSITIVITY AND SPECIFICITY: 99 percent.

LIMITATIONS: Diagnostic errors can occur due to rare sequence variations. F8 mutations, other than the F8 intron 22-A and intron 1 inversions, will not be detected.

Test developed and characteristics determined by ARUP Laboratories. See Compliance Statement C: aruplab.com/CS

H=High, L=Low, *=Abnormal, C=Critical

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
Maternal Contamination Study Fetal Spec	18-193-402149	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Maternal Contam Study, Maternal Spec	18-193-402149	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
F8 INV FE Specimen	18-193-402149	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Hemophilia A (F8) Inversions Interp	18-193-402149	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00

END OF CHART

H=High, L=Low, *=Abnormal, C=Critical